NCBDDDannualreport2011
National Center on Birth Defects and Developmental Disabilities • Centers for Disease Control and Prevention

Newborn Screening

Enhancing the Quality and Usefulness of Newborn Screening Data and Programs

What is the problem?

- In September 2011, HHS Secretary Sebelius approved adding critical congenital heart defects to the Recommended Uniform Screening Panel. Congenital heart defects (CHDs) account for 24% of infant deaths due to birth defects. In the United States, about 5,000 (or 12 per 10,000) babies born every year have critical congenital heart defects (CCHDs). Babies with a CCHD are at significant risk of disability or death if the condition is not diagnosed soon after birth. CCHDs can be detected in some babies using pulse oximetry, which is a test to determine the amount of oxygen in the blood.

- Long-term follow-up of children with confirmed newborn screening conditions ensures that these children receive the full benefits of early identification through newborn screening. Tracking these children is also important for public health. Efforts to systematically evaluate health outcomes, beyond long-term survival, with a few exceptions, are just beginning.

What do we know?

- In order to help states and health care providers as they implement the screening, it was recommended that the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children provide information to address the following issues:
    - What will be the impact on state health departments, including staffing needs, to implement this program? What are the roles of the state health departments?
    - What capability is present to ensure that all babies are screened and their results are communicated to providers, including assuring that those not screened at birth receive a screen?
- Technological improvements and partner collaboration have led to the expansion and increased uniformity of newborn screening as well as enhanced laboratory and data systems that provide better surveillance, tracking, and research.

- Each year, more than 4 million newborns in the U. S. are screened for hearing loss and certain genetic, endocrine, and metabolic disorders. Through early identification and treatment, newborn screening provides an opportunity for significant reductions in morbidity and mortality while reducing health care costs associated with treatment of lifelong debilitating conditions. Newborn screening not only saves lives, it can make lives healthier.

What can we do?

- As screening for critical congenital heart defects (CCHDs) is implemented throughout the U.S., CDC will play an important role in the surveillance and tracking of babies with a CCHD identified through CCHD screening using pulse oximetry. Evaluating the effectiveness of screening for CCHD will provide states and health care providers with data to better understand the potential impact of implementation and to help make decisions about adding this condition to their existing newborn screening program.

- CDC provides data and expertise toward development of state and national policies on newborn screening (NBS). This screening data are used to prevent or reduce the negative consequences of birth defects, developmental disabilities, and pediatric genetic conditions. To improve long-term follow-up, CDC develops methodology and tools to improve data collection processes and data quality for long-term follow-up after NBS.

- NCBDDD participated in the establishment and management of the Interagency Coordinating Committee (ICC) on Newborn and Child Screening outlined in the Newborn Screening Saves Lives Act of 2008. The ICC provides input from federal agencies to the Secretary of Health and Human Services regarding recommendations from the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children.

Did you know?

- A 2011 MMWR article included newborn screening as one of the Ten Significant Public Health Achievements — United States, 2001-2010, citing the improvements in technology and endorsement of a uniform newborn screening panel as leading to earlier life-saving treatment and intervention for newborns.

- The newborn screening system is comprehensive, including not only screening and diagnosis, but also long-term follow-up care through the health system. Long-term follow-up of affected children identified through newborn screening means that these children and their families will have the best outcomes.

Accomplishments

- NCBDDD showed that rates reported for certain newborn screening conditions have not changed substantially despite changes in laboratory technology. State newborn screening laboratories have used different methods and criteria for newborn dried blood spot screening. To assure that changes in protocol were not affecting the number of babies identified,

NCBDDD used data from the National Newborn Screening and Genetics Resource Center from 1991-2000 for congenital adrenal hyperplasia, phenylketonuria, and the sickle hemoglobinopathies. This study showed that there were no substantial changes in the birth prevalence rates of these conditions over the study period, which provided reassurance that changes in screening technologies over time did not negatively affect detection rates of diagnosed babies.

- NCBDDD has provided technical expertise and funded cooperative agreements with California, Iowa, New York, and Utah state health departments to incorporate newborn screening long-term follow-up into existing birth defects or newborn screening surveillance programs as part of a 3-year pilot project that began in September 2008. NCBDDD facilitates compilation and analysis of data from young children identified by newborn screening and disseminates methodology and results from multistate cooperative agreements to serve as a model for other state programs to conduct similar activities. Results will be available this year.

- NCBDDD collaborated with HRSA and the American Academy of Pediatrics (AAP) on a quality improvement project evaluating clinical support tools for newborn screening for use in the primary care setting. NCBDDD served on the Expert Group for the AAP Newborn Screen Positive Infant ACTion Project, in partnership with the American College of Medical Genetics, HRSA, and the AAP Quality Improvement Innovation Network. A summary of the project will be available this year. The Newborn Screen Positive Infant ACTion sheets were developed to guide primary care providers through short-term follow-up for newborns detected through public health screening for congenital conditions.

- NCBDDD published important considerations in the use of race and ethnicity to assess risk for certain blood disorders (hemoglobinopathies) in the newborn and prenatal settings. Certain blood disorders (hemoglobinopathies) differ among populations depending on their continent of origin, which is often thought of as race and ethnicity. Public health programs have debated the ethical and practical implications of targeted screening based on race/ethnicity versus screening everyone.

- NCBDDD worked with other federal agencies and partners to define and publish key questions that need to be addressed to achieve the long term follow-up goals of newborn screening, including care coordination, evidence-based treatment, continuous quality improvement, and new knowledge discovery.

Looking to the future

- NCBDDD has been tasked by the HHS Secretary to evaluate state surveillance and tracking systems to monitor the effectiveness of critical congenital heart defect (CCHD) newborn screening programs.

  - NCBDDD is assessing states' need for assistance with CCHD screening using pulse oximetry.
  - NCBDDD will continue to collaborate with New Jersey, the first state in the U.S. with legislatively mandated screening.
  - NCBDDD is working with New Jersey in early 2012 to assess surveillance and tracking of CCHD screening and to conduct a cost-effectiveness analysis of CCHD screening for the state.
  - NCBDDD is surveying birth defects surveillance programs to assess their potential roles with CCHD screening. The results of this survey will be published in 2012.

- NCBDDD has been tasked by the HHS Secretary to conduct a cost-effectiveness analysis of newborn screening, for the early identification of CCHDs.
  - NCBDDD is developing a cost-effectiveness model to identify the impact and cost of universal CCHD screening, including the average and incremental cost of CCHD screening per newborn, the number of infants with critical congenital heart defects identified through CCHD screening, the number of infant deaths avoided due to CCHD screening and the associated cost, and the number of life years gained due to CCHD screening and the associated cost.
- Subject matter experts from CDC are collaborating with the Association of Public Health Laboratories and members of the Subcommittee on Laboratory Standards and Procedures from the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children to evaluate the utility of routine second testing in newborn screening for the endocrinopathies (congenital hypothyroidism (CH) and congenital adrenal hyperplasia (CAH)).
- NCBDDD is working with the American Academy of Pediatrics to develop a training module on newborn screening to educate pediatricians about developing quality measures to ensure screening results are received and communicated to families. It will also provide approaches to working with specialty providers to coordinate care for children who have positive screening results. This effort could lead to improved follow-up care for children affected by newborn screening conditions.

Notable 2011 NCBDDD Scientifc Publications

- Hinton CF, Feuchtbaum L, Kus CA, Kemper AR, Berry SA, Levy-Fisch J, Luedtke J, Kaye C, Boyle CA. **What questions should newborn screening long-term follow-up be able to answer?: A statement of the United States Secretary for Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children**. Genetics in Medicine; 2011; 13:861-865.

- Hertzberg VS, Hinton CF, Therrell BF, Shapira SK. **Birth prevalence rates of newborn screening disorders in relation to screening practices in the United States**. Journal of Pediatrics; 2011; 159:555-560.
- Fisk Green R, Ehrhardt J, Ruttenber M, Olney RS. **Use of family history information for neural tube defect prevention: integration into state-based recurrence prevention programs**. American Journal of Health Education; 2011; 42: 296-308.
- Crider KS, Zhu J-H, Hao L, Yang Q-H, Gindler J, Maneval DR, Yang TP, Quinlivan EP, Li Z, Bailey LB, Berry RJ. **Methylenetetrahydrofolate reductase 677C→T genotype is associated with folate and homocysteine concentration in a large population-based double-blind trial of folic acid supplementation**. American Journal of Clinical Nutrition; 2011; 93:1365-1372.
- Hinton CF, Grant A, Grosse SD. **Ethical implications and practical considerations of ethnically-targeted screening for genetic disorders: the case of hemoglobinopathy screening**. Ethnicity and Health; 2011; 16:377-388.